Sara Nuovo

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7831670/publications.pdf

Version: 2024-02-01

19	382	933447	839539
papers	citations	10 h-index	18 g-index

20	20	20	795
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	1.2	66
2	Two unique TUBB3 mutations cause both CFEOM3 and malformations of cortical development. American Journal of Medical Genetics, Part A, 2016, 170, 297-305.	1.2	51
3	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. American Journal of Human Genetics, 2017, 101, 552-563.	6.2	45
4	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	4.5	36
5	Age and sex prevalence estimate of Joubert syndrome in Italy. Neurology, 2020, 94, e797-e801.	1.1	26
6	The spectrum of brainstem malformations associated to mutations of the tubulin genes family: MRI and DTI analysis. European Radiology, 2019, 29, 770-782.	4.5	22
7	SUFU haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. Journal of Medical Genetics, 2022, 59, 888-894.	3.2	19
8	Between SCA5 and SCAR14: delineation of the SPTBN2 p.R480W-associated phenotype. European Journal of Human Genetics, 2018, 26, 928-929.	2.8	17
9	A novel IRF2BPL truncating variant is associated with endolysosomal storage. Molecular Biology Reports, 2020, 47, 711-714.	2.3	16
10	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. Nephrology Dialysis Transplantation, 2020, 35, 1195-1202.	0.7	15
11	Refining the mutational spectrum and geneâ€“phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. Journal of Medical Genetics, 2022, 59, 399-409.	3.2	13
12	Movement disorders and brain iron overload in a new subtype of aceruloplasminemia. Parkinsonism and Related Disorders, 2015, 21, 658-660.	2.2	10
13	Targeted Next Generation Sequencing in patients with Myotonia Congenita. Clinica Chimica Acta, 2017, 470, 1-7.	1.1	10
14	The Use of New Mobile and Gaming Technologies for the Assessment and Rehabilitation of People with Ataxia: a Systematic Review and Meta-analysis. Cerebellum, 2020, 20, 361-373.	2.5	10
15	Clinical variability at the mild end of BRAT1 â€related spectrum: Evidence from two families with genotypeâ€“phenotype discordance. Human Mutation, 2022, 43, 67-73.	2.5	9
16	CASK related disorder: Epilepsy and developmental outcome. European Journal of Paediatric Neurology, 2021, 31, 61-69.	1.6	7
17	Genetics of cerebellar disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 154, 267-286.	1.8	3
18	Novel unconventional variants expand the allelic spectrum of OPHN1 gene. American Journal of Medical Genetics, Part A, 2021, 185, 1575-1581.	1.2	3

#	Article	IF	Citations
19	Challenges and resources in adult life with Joubert syndrome: issues from an international classification of functioning (ICF) perspective. Disability and Rehabilitation, 2021, , 1-8.	1.8	1